



CCN6 gene

cellular communication network factor 6

Normal Function

The *CCN6* gene provides instructions for making a protein that appears to be involved in bone growth and the maintenance of cartilage, which covers and protects the ends of bones. The function of the CCN6 protein is not well understood. It is part of a family of proteins that are involved in the growth and maintenance of connective tissues, such as bone, cartilage, and blood vessels. The CCN6 protein is made in cells called chondrocytes, which produce and maintain cartilage, and is associated with the production of certain proteins that make up cartilage, but its role in their production is unclear. CCN6 may also help control signaling pathways involved in the development of cartilage and bone and may help regulate the breakdown of cartilage components.

Health Conditions Related to Genetic Changes

Progressive pseudorheumatoid dysplasia

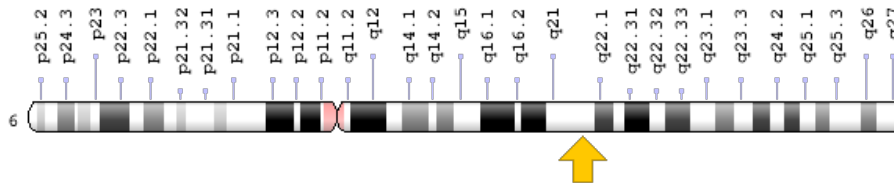
Mutations in the *CCN6* gene cause progressive pseudorheumatoid dysplasia (PPRD), which is a condition that causes stiffness and pain in the joints of the hands, hips, knees, and spine. The joint problems worsen over time, and movement in the joints becomes limited. Most of the mutations involved in this condition lead to production of an abnormally short CCN6 protein that is probably nonfunctional. Other mutations change single protein building blocks (amino acids) in the protein. Loss of CCN6 protein function likely disrupts normal cartilage maintenance and bone growth, leading to the joint problems in PPRD.

Juvenile idiopathic arthritis

Chromosomal Location

Cytogenetic Location: 6q21, which is the long (q) arm of chromosome 6 at position 21

Molecular Location: base pairs 112,052,813 to 112,069,686 on chromosome 6 (Homo sapiens Updated Annotation Release 109.20200522, GRCh38.p13) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- CCN family member 6
- LIBC
- PPAC
- PPD
- WISP-3
- WISP3
- WISP3_HUMAN
- WNT1 inducible signaling pathway protein 3
- WNT1-inducible-signaling pathway protein 3

Additional Information & Resources

Clinical Information from GeneReviews

- Progressive Pseudorheumatoid Dysplasia
<https://www.ncbi.nlm.nih.gov/books/NBK327267>

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28WISP3%5BTIAB%5D%29+OR+%28WNT1+inducible+signaling+pathway+protein+3%5BTIAB%5D%29+OR+%28CCN6%29%29+OR+%28%28WNT1-inducible-signaling+pathway+protein+3%5BTIAB%5D%29+OR+%28WISP-3%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D>

Catalog of Genes and Diseases from OMIM

- WNT1-INDUCIBLE SIGNALING PATHWAY PROTEIN 3
<http://omim.org/entry/603400>

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
<http://atlasgeneticsoncology.org/Genes/WISP3ID469ch6q22.html>
- ClinVar
<https://www.ncbi.nlm.nih.gov/clinvar?term=CCN6%5Bgene%5D>
- HGNC Gene Symbol Report
https://www.genenames.org/data/gene-symbol-report/#!/hgnc_id/HGNC:12771
- Monarch Initiative
<https://monarchinitiative.org/gene/NCBIGene:8838>
- NCBI Gene
<https://www.ncbi.nlm.nih.gov/gene/8838>
- UniProt
<https://www.uniprot.org/uniprot/O95389>

Sources for This Summary

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 - OMIM: WNT1-INDUCIBLE SIGNALING PATHWAY PROTEIN 3
<http://omim.org/entry/603400>
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